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Supravalvular aortic stenosis
Echocardiographic features

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The echocardiographic manifestations of segmental supravalvular aortic stenosis are described in 2 patients. The diagnosis was confirmed by cardiac catheterization in both and at operation in 1. A systematic echocardiographic approach to such patients is described. The characteristic finding in these patients was the narrowing of the diameter of the aortic lumen at the stenotic area just distal to the aortic valve. As the transducer sweeps further cephalad the aortic lumen widens to a normal diameter. In one patient treated surgically, postoperative echogram demonstrated the narrowing to be reduced.

Echocardiography has been well established as a noninvasive procedure in the diagnosis of disease processes involving the left ventricular outflow tract. Since Edler *et al.*'s (1961) original echocardiographic description of the aortic valve, abnormal aortic valve echo patterns have been described in such conditions as valvular aortic stenosis (Gramiak and Shah, 1970; Feizi, Symons, and Yacoub, 1974), hypertrophic muscular (Shah *et al.*, 1971) as well as discrete membranous subaortic stenosis (Popp *et al.*, 1974; Davis *et al.*, 1974), truncus arteriosus (Feigenbaum, 1972; Chung *et al.*, 1973), bicuspid aortic valve (Nanda *et al.*, 1974), endocarditis (Dillon *et al.*, 1973), prosthetic aortic valves (Douglas and Williams, 1974), aortic root dissection (Nanda, Gramiak, and Shah, 1973), and right sinus of Valsalva aneurysm (Rothbaum *et al.*, 1974). However, at the time of writing there is only one recently published case report of an echocardiogram in a patient with supravalvular aortic stenosis (Usher, Goulden, and Murgo, 1974). In the present report we describe two patients with clinical, haemodynamic, angiographic, and anatomical features of supravalvular aortic stenosis who had distinctive echocardiographic features.

Methods

Echocardiography was performed using a Unirad ultrasonoscope with a 2.25 MHz transducer, 1.3 cm in diameter, having an acoustic lens providing beam collimation at 5 cm tissue depth. The echocardiograms were recorded using a Polaroid camera. In Case 2, a strip chart record (Honeywell No. 1856 Fibroptics recorder) was obtained during the postoperative evaluation. The patients were studied in the supine position, with the transducer placed in the third or fourth intercostal space in order to record the free edge of the mitral valve with the transducer oriented perpendicular to the chest wall. The transducer was directed cephalo-medially from the mitral valve to the aortic valve and aortic root, while sequential Polaroid tracings or continuous strip chart recordings were obtained.

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Case reports

Case 1

A 9-year-old girl was referred to the Texas Children's Hospital, Houston, Texas, with a heart murmur and fainting episodes of three weeks' duration. The newborn period was uncomplicated and she had an apparently normal development. A routine examination was first made in October 1973 when a heart murmur and an arrhythmia were noted. During the month preceding admission she experienced dyspnoea on exertion and fainting spells while standing in line at school. There was no history of febrile illness or toxic exposure of the mother during pregnancy. Her mother and a paternal uncle were reported to have congenital aortic stenosis but there was no similar condition in her sib. Her father had no clinical evidence of heart disease.

Physical examination at the time of admission revealed a well-developed girl in no distress. Blood pressure, pulse rate, and respiratory rate were: 120/80 mmHg (16.0/10.6 kPa), 100 beats/min, and 20 respirations/min, respectively. A diminished upstroke of the

brachial pulse was evident. Fine crepitant râles were audible over the lower posterior lung fields. A systolic thrill was palpable in the suprasternal notch and over the carotid arteries. A sustained apical impulse was located at the 5th intercostal space, anterior axillary line. The first heart sound was slightly diminished in intensity; the second heart sound was closely but physiologically split. There were no clicks or gallops. A grade 5/6 harsh systolic ejection murmur was best heard along the right sternal border in the second intercostal space but was also heard at the left sternal border, suprasternal notch, apex, and over the carotids. The remainder of the examination was normal.

Laboratory studies including a blood picture, urinalysis, electrolytes, and serum calcium were normal. A chest *x*-ray revealed cardiomegaly. The electrocardiogram demonstrated sinus rhythm, and left ventricular hypertrophy with left ventricular strain pattern.

The echocardiogram from this patient is illustrated in Fig. 1. Scanning through the ascending aorta showed a significant narrowing of the aortic root distal to the aortic cusps. The internal diameter of the stenotic segment measured 0.47 cm compared to 1.64 cm diameter of the aorta at the valve level. As the sweep of the transducer was continued cephalad the aortic root widened to 1.32 cm. The echographic pattern was quite reproducible.

Left and right heart catheterization was performed and the haemodynamic data are summarized in the Table. A gradient of 100 mmHg (13.3 kPa) across the aortic valve was noted. Damping of the pressure curve occurred as the catheter was withdrawn from the aortic valve area to the ascending aorta and a 80 mmHg (10.6 kPa) peak systolic gradient was recorded across the stenotic area. The stenotic supravalvular area was confirmed by left ventricular cineangiography (Fig. 2). The ratio of the stenotic segment to the post-stenotic internal diameter was 0.39 by angiography and 0.36 (0.47/1.32) by echography. Right ventricular hypertrophy, infundibular pulmonary stenosis, and bilateral pulmonary arterial branch stenosis were also found by right ventricular catheterization and angiography.

At operation, there was extensive hypertrophy of the left ventricle and some hypertrophy of the right ventricle. Stenosis of the aortic valve with fusion of the right and non-coronary cusps was observed. The aortic wall was thickened for a length of about 3 cm starting just distal to the left coronary cusp. Wedge resection of the stenotic area and a 3×1.5 cm woven 'dacron' patch angioplasty were performed. A commissurotomy between the right and non-coronary cusps was also done. Postoperatively the left ventricular systolic pressure was 175 mmHg (23.3 kPa), and aortic root systolic pressure above the patch was 140 mmHg (18.6 kPa). Her postoperative course was uneventful. Histology of the aortic wall

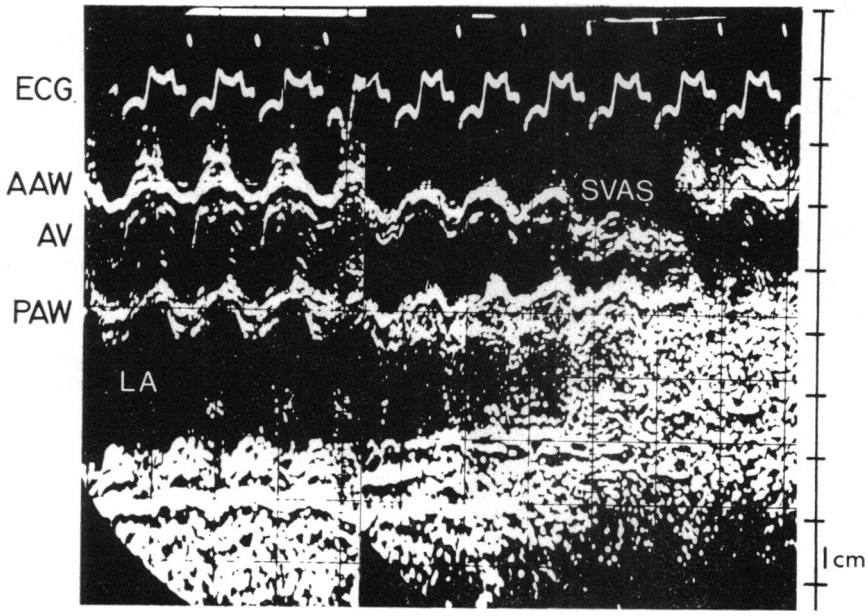

FIG. 1 *Preoperative echocardiogram from Case 1. The transducer sweeps cephalad from the aortic root. As the echoes of the aortic valve cusp disappear, narrowing of the aortic root occurs. Increased echoes are evident at the site of the stenosis. As the sweep of the transducer continues cephalad the aortic root widens. Abbreviations: AAW = anterior aortic wall; AV = aortic valve; PAW = posterior aortic wall; LA = left atrium; SVAS = supravalvular aortic stenosis. Polaroid pictures fused together for the purpose of illustration.*

TABLE *Haemodynamic data in Cases 1 and 2*

	Case 1 *Pressure* *(mmHg)* *Phasic*	*Mean*	*Case 2* *Pressure* *(mmHg)* *Phasic*	*Mean*
Right atrium	a=4; v=2 *a=0.5; v=0.3*	3 *0.4*	a=4; v=5 *a=0.5; v=0.7*	4 *0.5*
Right ventricle	80/0–5 *10.6/0–0.7*	—	38/0–5 *5.1/0–0.7*	—
Right ventricular outflow	50/0–8 *6.7/0–1.1*	—	28/0–5 *3.7/0–0.7*	—
Main pulmonary artery	50/8 *6.7/1.1*	17 *2.3*	28/10 *3.7/1.3*	17 *2.3*
Right pulmonary artery	16/4 *2.1/0.5*	12 *1.6*	22/9 *2.9/1.2*	14 *1.9*
Left pulmonary artery	16/8 *2.1/1.1*	12 *1.6*	20/10 *2.7/1.3*	13 *1.7*
Pulmonary wedge	—	8 *1.1*	a=12; v=8 *a=1.6; v=1.1*	9 *1.2*
Left ventricle	280/0–8 *37.2/0–1.1*	—	190/0–9 *25.3/0–1.2*	—
Ascending aorta distal to valve	180/70 *23.9/9.3*	112 *14.9*	170/70 *22.6/9.3*	—
Ascending aorta distal to stenosis	100/70 *13.3/9.3*	100 *13.3*	120/70 *16.0/9.3*	90 *12.0*

Note: Figures in italics are pressures in kPa.

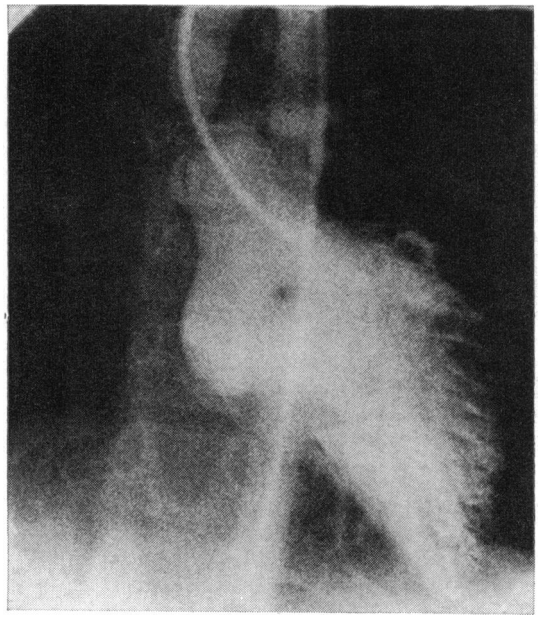

FIG. 2 *Frame of left ventricular angiogram from Case 1 in the anteroposterior projection showing a narrowed area just above the coronary ostia and a normal size aorta distal to the stenosis. The aortic valve leaflets are irregularly thickened and are dome shaped during systole. The left coronary artery is dilated.*

biopsy disclosed medial hypertrophy with medial degeneration and mucopolysaccharide deposits. An echocardiogram was obtained on the eighth postoperative day and a supravalvular aortic narrowing was no longer visualized (Fig. 3).

Case 2

A 7-year-old boy was admitted to Texas Children's Hospital, Houston, Texas, on 2 June 1974 because of fever, toothache, and irritability of a few days' duration. At the age of 3 months a heart murmur was noted on physical examination. The diagnosis of valvular (bicuspid) and supravalvular aortic stenosis was made during cardiac catheterization at the age of 5 years, and an aortic valve commissurotomy and 'dacron' patch angioplasty was performed. His postoperative course was complicated by antero-septal myocardial infarction from which he completely recovered.

Physical examination on admission revealed a thin boy with obvious microphthalmia on the right; the eye appeared amblyopic. His temperature was normal and his pulse rate was 104 beats per minute with normal peripheral pulses. The blood pressure in the right arm and right leg were 110/70 and 120/80 mmHg (14.6/9.3 and 16.0/10.6 kPa), respectively. There was no venous distension but a prominent suprasternal notch pulsation was present. The cardiac apical impulse was located in the fifth intercostal space 1 cm lateral to the midclavicular line. A systolic thrill was felt all over the praecordium and over the carotid arteries. On auscultation, the first heart sound was normal. A grade 4/6 holosystolic murmur was heard over the praecordium, carotids, and axillae. The second heart sound was reduced and obscured by

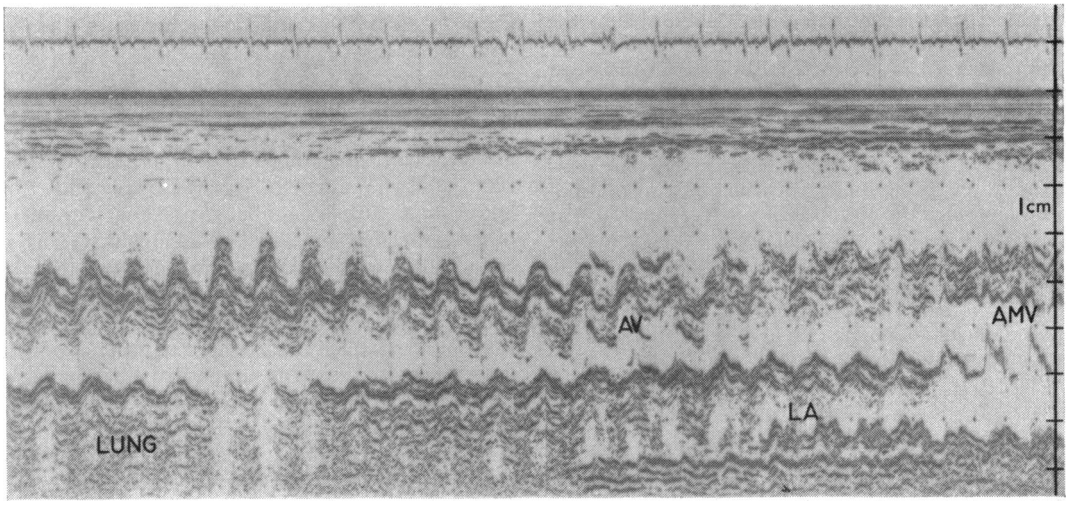

FIG. 3 *Postoperative continuous strip chart recording of echocardiogram from Case 1. The transducer sweeps inferiorly from the ascending aorta, to aortic valve, and mitral valve. The area of supravalvular stenosis is not apparent. Abbreviations: AMV = anterior mitral valve leaflet. The remainder are as in Fig. 1.*

the systolic murmur. A grade 1/4 early diastolic blowing murmur was heard over the left sternal border. Other physical findings were unremarkable. Laboratory studies including haemogram, urinalysis, electrolytes, and serum calcium were normal. There was no clinical or laboratory sign of subacute bacterial endocarditis.

A chest *x*-ray was normal. The electrocardiogram revealed normal sinus rhythm, deep inverted T waves, and poor R wave progression in the anterior praecordial leads.

The echocardiogram from this patient is illustrated in Fig. 4. Scanning through the ascending aorta showed a

FIG. 4 *Echocardiogram from Case 2. As the transducer sweeps cephalad from the aortic root, aortic valve cusps disappear, and narrowing of the aortic root occurs. As the sweep of the transducer continues cephalad the aortic root widens. Notice the eccentricity of the aortic valve echo during diastole indicative of bicuspid aortic valve. Abbreviations: same as Fig. 1. Polaroid pictures fused together for illustrative purposes.*

significant narrowing of the aortic root distal to the aortic cusps. The internal diameter of the stenotic segment measured 0.93 cm compared to 1.6 cm diameter of the aorta at the valve level. As the sweep of the transducer was continued cephalad the aortic root widened to 1.6 cm. The patient subsequently underwent heart catheterization, and the haemodynamic data are summarized in the Table. Left ventricular cineangiocardiography confirmed the diagnosis of residual supravalvular stenosis (peak gradient of 50 mmHg (6.7 kPa)) above the previously placed 'dacron' patch angioplasty (Fig. 5). The ratio of the stenotic segment to the proximal internal diameter was 0.50 by angiography and 0.58 by echography. A small 20 mmHg (2.7 kPa) gradient across the aortic valve was also observed.

Discussion

Supravalvular aortic stenosis may assume one of three forms, namely (1) discrete membranous (2) the hour-glass or segmental, and (3) the diffuse hypoplastic (Peterson, Todd, and Edwards, 1965). The hour-glass form is the most frequent, and it is characterized by segmental involvement of the ascending aorta with thickening of the wall of the aorta, resulting in a narrow lumen, while the external diameter of the vessel may either be normal or slightly narrow. This type of supravalvular aortic stenosis may be associated with similar changes in the pulmonary arteries (as was demonstrated in Case 1) as well as in the branches of the aorta (Blieden *et al.*, 1974).

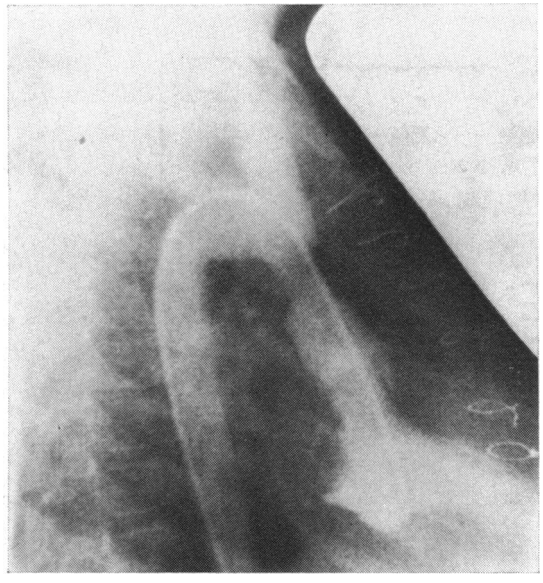

FIG. 5 *Frame of left ventricular cineangiogram after operation from Case 2 in 60° right anterior oblique projection showing a narrowed segmental area before the bifurcation of the ascending aorta.*

Supravalvular aortic stenosis has been described in association with the idiopathic hypercalcaemia syndrome of infancy (Black and Bonham Carter, 1963), with or without elfin facies or mental retardation (Williams, Barratt-Boyes, and Lowe, 1961), and with a familial syndrome without any other associated anomalies (McDonald, Gerlis, and Somerville, 1969; Martin and Moseley, 1973). It would, therefore, be useful to have a noninvasive, nontraumatic technique, like echocardiography, that could establish the presence or absence of supravalvular aortic stenosis in patients having any of these syndromes which may be associated with this congenital abnormality. Recognition of supravalvular aortic stenosis by echocardiography could also be used to gain further knowledge on the familial incidence of the disease.

Supravalvular aortic stenosis may closely mimic the usual type of valvular or subvalvular aortic stenosis in all aspects of the history, physical examination, chest *x*-ray, and electrocardiograms. The absence of an ejection click may be the most consistently reliable clue to differentiate supravalvular and subvalvular from aortic valvular stenosis (Popp *et al.*, 1974). Echocardiographic criteria for the diagnosis of discrete membranous and hypertrophic muscular subaortic stenosis have been well established (Shah *et al.*, 1971; Popp *et al.*, 1974; Davis *et al.*, 1974). Though echocardiographic findings in valvular aortic stenosis have been described, their clinical applications are limited by their lack of specificity in differentiating valvular stenosis from insufficiency (Gramiak and Shah, 1970; Feigenbaum, 1972). In addition, valvular aortic stenosis may coexist with supravalvular stenosis (as seen in our two patients), as well as with subaortic membranous or muscular stenosis (Parker, Kaplan, and Connolly, 1969). Recently, Nanda *et al.* (1974) have described echocardiographic criteria for diagnosis of congenital bicuspid valve. The aortic valve echogram from Case 2 showed an eccentricity index of 2.0; a bicuspid aortic valve was diagnosed angiographically and by surgery at age 5.

Our 2 cases of segmental supravalvular aortic stenosis as well as another case report (Usher *et al.*, 1974) demonstrated a distinct echographic abnormality which correlated well with the angiographic and anatomical features of the disease. The good correlation observed in measuring the percentage narrowing of the stenotic segment between echocardiography and angiography in our 2 cases suggests that the echocardiogram may also prove helpful in estimating the degree of severity of the stenosis. Experience with a larger series of cases will

be needed to determine the full potential of this noninvasive technique in the assessment of supravalvular stenosis.

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